Award Number: W81XWH-13-2-0010

TITLE: Semantic Web Technology for Mapping and Applying Clinical Functional Assessment Information

PRINCIPAL INVESTIGATOR: Mark A. Musen, MD PhD

CONTRACTING ORGANIZATION: Stanford University

Menlo Park, CA 94025-3434

REPORT DATE: May 2015

TYPE OF REPORT: Final

PREPARED FOR: U.S. Army Medical Research and Materiel Command

Fort Detrick, Maryland 21702-5012

DISTRIBUTION STATEMENT: Approved for Public Release; Distribution Unlimited

The views, opinions and/or findings contained in this report are those of the author(s) and should not be construed as an official Department of the Army position, policy or decision unless so designated by other documentation.

# REPORT DOCUMENTATION PAGE

Public reporting burden for this collection of information is estimated to average 1 hour per response, including the time for reviewing instructions, searching existing data sources, gathering and maintaining the data needed, and completing and reviewing this collection of information. Send comments regarding this burden estimate or any other aspect of this collection of information, including suggestions for reducing this burden to Department of Defense, Washington Headquarters Services, Directorate for Information Operations and Reports (0704-0188), 1215 Jefferson Davis Highway, Suite 1204, Arlington, VA 22202-4302. Respondents should be aware that notwithstanding any other provision of law, no person shall be subject to any penalty for failing to comply with a collection of information if it does not display a currently valid OMB control number. PLEASE DO NOT RETURN YOUR FORM TO THE ABOVE ADDRESS.

Form Approved

OMB No. 0704-0188

1. REPORT DATE	2. REPORT TYPE	3. DATES COVERED					
May 2015	Final	15Feb2013 - 14Feb2015					
4. TITLE AND SUBTITLE		5a. CONTRACT NUMBER					
Semantic Web Technology for Map	W81XWH-13-2-0010						
<b>Functional Assessment Information</b>		5b. GRANT NUMBER					
		5c. PROGRAM ELEMENT NUMBER					
6. AUTHOR(S)		5d. PROJECT NUMBER					
Mark A. Musen MD PhD							
		5e. TASK NUMBER					
	5f. WORK UNIT NUMBER						
E/Mail: musen@stanford.edu							
7. PERFORMING ORGANIZATION NAME(S	S) AND ADDRESS(ES)	8. PERFORMING ORGANIZATION REPORT NUMBER					
Stanford University							
Menlo Park, CA 94025-3434							
9. SPONSORING / MONITORING AGENCY	NAME(S) AND ADDRESS(ES)	10. SPONSOR/MONITOR'S ACRONYM(S)					
U.S. Army Medical Research and M	lateriel Command						
Fort Detrick, Maryland 21702-5012							
-		11. SPONSOR/MONITOR'S REPORT					
		NUMBER(S)					
40 DIGEDIDITION / AVAIL ADULTY OF A TO		1					

#### 12. DISTRIBUTION / AVAILABILITY STATEMENT

Approved for Public Release; Distribution Unlimited

## 13. SUPPLEMENTARY NOTES

#### 14. ABSTRACT

Our project analyzed retention standards, Disability Benefit Questionnaires (DBQs), the Military Occupation Specialties (MOS) manual, and the Department of Veteran Affairs schedule for rating disabilities, and it developed a novel framework for structuring and using functional assessment information. Within this framework, our team modeled as OWL ontologies descriptions of functional assessments and their value sets (CFA ontologies). We created models for patient-specific functional-assessment data and for assessment forms such as DBQs. We mapped the functional assessments descriptors in our CFA ontologies to categories and qualifiers in the International Classification of Functioning, Disability, and Health (ICF). We developed the mechanisms to generate programmatically data-acquisition Web forms from the CFA ontologies and we demonstrated how a clinician user might use the forms to document clinical functional assessment and to generate structured data conformant with the CFA ontologies as part of the process. Backend software transforms the structured data to tab-delimited files and to RDF and OWL individuals. We developed mechanisms to link the data with CFA ontologies and to make the data queryable. The resulting software thus provides an end-to-end means to model patient data related to functional assessment, to acquire such data from clinicians, and to output the data in a form suitable for further processing

#### 15. SUBJECT TERMS

Functional Status; International Classification of Functioning, Disability, and Health; Disability Benefit Questionnaire; Integrated Disability Evaluation System

16. SECURITY CLAS	SSIFICATION OF:		17. LIMITATION OF ABSTRACT	18. NUMBER OF PAGES	19a. NAME OF RESPONSIBLE PERSON USAMRMC
a. REPORT U	b. ABSTRACT U	c. THIS PAGE U	υυ	32	19b. TELEPHONE NUMBER (include area code)

## **Table of Contents**

	<u>P</u>	age
1	Introduction	1
2.	Keywords	4
3.	Overall Project Summary	4
4.	Key Research Accomplishments	. 17
5.	Conclusion	. 18
6.	Publications, Abstracts, and Presentations	. 18
7.	Inventions, Patents, and Licenses	. 18
8.	Reportable Outcomes	. 18
9.	Other Achievements	. 19
10.	References	. 19
11.	Participants	. 19
12.	Annendices	. 19

## 1. Introduction

In early 2013, the Stanford Center for Biomedical Informatics Research (BMIR) received a contract from TATRC to develop a "Common Language" for clinical functional assessment (CFA-CL). It was a two-year contract starting in February 2013 and terminating in February 2015. This document is the final report that summarizes results obtained in the two-year project. It describes our findings on the DoD/VA's Integrated Disability Evaluation System (IDES), our revised project goals, our modeling approach, and the artifacts developed in the project. To make this report self-contained, when appropriate, we incorporate the findings reported in the annual report submitted at the end of the first contract year. We submit as appendices two papers, one titled "Structured Data Acquisition with Ontology-Based Web Forms" and the second titled "Driving Structured Data Entry for Functional Assessment Using Standard Terminologies." As of this writing, the former paper has been accepted for publication and presentation at the International Conference on Biomedical Ontology 2015 (Lisbon, Portugal). The latter paper is under review for presentation at the American Medical Informatics Association 2015 Annual Symposium.

## 2. Keywords

Functional Status; International Classification of Functioning, Disability, and Health; Disability Benefit Questionnaire; Integrated Disability Evaluation System

## 3. Overall Project Summary

We have named the project "FACSIMILE." Its initial goals, as described in our original proposal, include:

- Development of a "Common Language" (CFA-CL) for clinical function assessments that is grounded in International Classification of Functioning, Disability, and Health (ICF)
- Demonstration that data used in DoD/VA's Integrated Disability Evaluation System (IDES) can be coded in this common language
- Demonstration of uses of coded clinical function assessment data in the IDES process
- Creation of a prototype CFA semantic model in which categories of impairment are defined by constraint expressions consisting of the CFA-CL and ICF code stems, qualifiers, and qualifier values.

Our focus has been IDES, through which DoD and VA providers and coordinators both evaluate a service member for fitness for service and determine a possible disability rating in parallel, thus reducing the required processing time for a disabled service member to begin receiving benefits. In the following, we describe the results of the project in terms of the tasks outlined in the Statement of Work.

# 1. Analyze the functional requirements of tasks in the Integrated Disability Evaluation System (IDES) workflow where clinical functions are assessed, documented, stored, transmitted, and used.

In the first part of this project, we engaged in extensive consultation with colleagues in Madigan Army Medical Center to determine (1) the nature of clinical assessment information generated and used in IDES, and (2) opportunities to use coded clinical functional assessment information to inform decision-making in IDES.

In the IDES process, when the illness or injury of a service member fits the criteria defined in the medical fitness standards for retention and separation (e.g., Army Regulation 40-501[2]), and further treatment will not cause the member to meet medical retention standards or render them capable of performing the duties required by their office, grade, rank, and rating, the heath-care provider refers the service member to a Medical Evaluation Board (MEB) for the initiation of the IDES process and a Physical Evaluation Board Liaison Officer (PEBLO) is appointed for the service member. The PEBLO prepares and submits the case file to a VA Military Service Coordinator (MSC), who initiates VA processing of the case, schedules a medical exam, and sends the exam results to the PEBLO. The MEB providers use all available information to produce a narrative summary. The narrative summary, together with the service member's medical and service profiles and the history and

<sup>&</sup>lt;sup>1</sup> Functional-Assessment Coding for Semantic Interpretation of Military Impairment-Level Evaluation

treatment of the injury or illness, is used by the MEB to determine whether the member has a medical condition that is incompatible with continued military service in his or her current capacity. After a review process, the case file is sent to the Physical Evaluation Board (PEB) for determining the service member's fitness for service. An informal PEB makes the initial determination, and if the service member is found unfit, submits a request for disability ratings of all claimed conditions. A Disability Rating Activity Site issues a rating based on the findings of the VA medical examination. The process ends when all reviews and appeals have been processed and the disposition of the case is approved by the Physical Disability Agency (PDA) for the service member's return to duty or for the issuance of a VA's benefits decision letter.

We found that Madigan AMC's IDES data processing relies on PDF documents. The documents include narrative summaries in free text (Figure 1) or form-based documents that are accessible as PDFs (Figure 2). Because there is no coding scheme for clinical functional assessments (something that this project aimed to address), functional assessment information in Madigan's IDES documentation is scattered in various narrative documents. ICD codes are the only structured data that are readily available. We procured an example of the dossier that is generated for a service member. It consisted of 45 pages of mostly narrative notes that would require significant time to redact and de-identify. The dossier provided many examples of clinical functional assessments (e.g., see highlighted text in Figure 1). However, it would take herculean effort to convert such free text into coded data post hoc. Within the workflow of IDES as carried out at Madigan, we saw no prospect of such structured coding being done. We concluded that it was unrealistic to expect that we could obtain a large sample of de-identified data from Madigan.

Furthermore, it was not clear what would be a good use case for the structured functional assessment information in Madigan's current IDES process. Madigan MEB physicians and a PEB officer emphasized to us in interviews that the retention decision is based on a holistic evaluation of many sources of information, including the service member's motivation and his/her superiors' assessments, rather than on any kind of structured assessment data. We struggled to find decision points where structured data could play a role in Madigan's IDES process.

Nevertheless, IDES is a complex and evolving process where a number of DoD and VA information systems interact with each other. We did not rule out the possibility of structured functional assessment data becoming useful in Madigan's IDES process in the future.

## SPECIFIC HISTORY FOR: CHRONIC BILATERAL HIP CONDITION SECONDARY TO OSTEOARTHRITIS

The claimant reports being diagnosed with CHRONIC BILATERAL HIP CONDITION SECONDARY TO OSTEOARTHRITIS'. The condition has existed since 2005. The condition started after a 12 mile road march He reports the following symptom(s): pain. He indicates he does not experience weakness, stiffness, swelling, heat, redness, giving way, lack of endurance, locking, fatigability, deformity, tenderness, drainage, effusion, subluxation and dislocation. The claimant reports experiencing the following flare-ups as often as 7 time(s) per week and each time lasts for 1 day(s). From 1 to 10 (10 being the worst) the severity level is at 10. The flare-ups are occurring spontaneously. It is alleviated by rest, spontaneously and by Toradol. During the flare-ups he experiences the following functional impairment which is described as not able to run or walk fast and limitation of motion of the joint which is described as limited range of motion. He reports difficulty with standing/walking. The claimant describes can't stand or sit still for more than 30 minutes The treatment is Toradol, physical training, Tylenol

Figure 1. Example of functional assessment information embedded in narrative text of notes.

For use of this form, see	AR 40~		SICAL PROFILE	Office	of the	Surge	on General.					
	For use of this form, see AR 40-501; the proponent agency is the Office of the Surgeon General.  MEDICAL CONDITION: (Description in lay terminology) X INJURY? Or ILLNESS/DISEASE? 2. CODES (Table 3.							Р	Ų	L F	П	E S
Internal hip pain (femoral acotabular impingement left greater than right, and osteoarthritis); obstructive sleep apnea (								· 2	1	3	7	1 1
4. PROFILE TYPE					····					YES	I	NO
a. TEMPORARY PROFILE (Expiration date YYYYMMDD)			(Limited to 3 r.	nonths	duration	}					$\perp$	×
b. PERMANENT PROFILE (Reviewed and validated with e	very peri	odic healt	h assessment or after 5 years fr	am the	date of	issue)				×		
5. FUNCTIONAL ACTIVITIES THAT EVERY SOLDIER REC THESE TASKS, THEN THE PULHES MUST CONTAIN AT	GARDLI LEAST (	ESS OF ONE "3"	MOS MUST BE ABLE TO PE AND SOLDIER MUST BE R	ERFOR EFER	RM. IF RED TO	SOLDI A ME	ER CANNOT PER B. CAN THE SOLI	FORI	M AN	Y ONE	: OF	:
FUNCTIONAL ACTIVITY:								YES	-	ИО		
a. Carry and fire individual assigned weapon?										×		
b. Evade direct and indirect fire?											I	X
c. Ride in a military vehicle for at least 12 hours per day	1?									X	Τ	
d. Wear a helmet for at least 12 hours per day?							×	$\top$				
e. Wear body armor for at least 12 hours per day?										$\perp$	×	
f. Wear load bearing equipment (LBE) for at least 12 hours per day?									$\perp$	X		
g. Wear military boots and uniform for at least 12 hours per day?								X	$\top$			
h. Wear protective mask and MOPP 4 for at least 2 continuous hours per day?									×			
i. Move 40lbs (for example, duffie bag) while wearing usual protective gear (helmet, weapon, body armor and LBE) at least 100 yards?									X			
j. Live in an austere environment without worsening th	e medic	cal condi	tion?									X
6. APFT	YEŞ	NO	ALTERNATE APET (Fill or	ut if une	ble to d	o APFT	run otherwise N/A)	N/	Α	YES		NO
2 MILE RUN		X	APFT WALK								$\perp$	×
APFT SIT-UPS		×	APFT SWIM								$\perp$	×
APFT PUSH UPS	×		APFT BIKE			•					$\perp$	×
7. DOES THE SOLDIER MEET RETENTION STANDARDS	IAW CI	HAPTER	3 AR 40-501?									
YES NEEDS MMR	RB		№ 🗙	NEEL	DS ME	В						
8. FUNCTIONAL LIMITATIONS AND CAPABILITIES AND ON Crawling, Crouching, Running, or Weight bearing for multiple was to wearing individual body armor. No wearing rucksack, No	ore than	20 pour	nds.	mpact	activity	No sit	-ups. No flutter kic	ks. No	wall	king gr	eate	r

Figure 2. Example of formed based information collection.

Locally, we interviewed Dr. Michael Tierney, a physician at the VA Palo Alto Health Care System who evaluates service members from all branches of the military. These interviews revealed the variations in the IDES documentation practices of different service branches. In early 2013, for example, the Navy used Disability Benefit Questionnaires (DBQs), which are problem-specific assessment instruments whose component questions are designed to elicit the information needed to complete a disability rating based on the rating schedules of Code of Federal Regulations Part 4. In early 2013, the workflow in Army's IDES did not use DBQs. However, according to members of our Advisory Board, all branches subsequently have transitioned to the use of DBQs. The current Separation Health Assessment (SHA) makes use of a General Medical (Gen Med) Examination DBQ template, which is intended to be a brief clinical summary and which requires that the details of each condition to be recorded in the individual specialty DBQs.

In response to these developments, we focused our attention on DBQs as potential instruments for capturing structured functional assessment information. We developed semantic models of typical DBQs, investigated the nature of data elements in DBQs, developed CFA-CL that models the semantics of DBQ data elements, and showed how such model can drive the generation of forms to acquire such structured data and how such data can be queried.

## 2. Propose a structure for CFA-CL. The CFA-CL coding scheme will be described in a document and also modeled as an OWL ontology using the Protégé tool.

In our original proposal, we hypothesized that CFA-CL codes will have the form of NNNN.e.xxxx, where *NNNN* is an ICF stem code at either the 3 or 4 digit level, *e* is an optional extension code that augments the ICF code to have greater specificity than that which is available in ICF, and *xxxx* denotes a set of category-specific qualifiers. For example, the *Hip and Thigh Conditions* DBQ evaluates the function of the hip in terms flexion, extension, abduction, and rotation. In ICF, the closest code for these functions is b7100 (functions of the range

and ease of movement of one joint). We hypothesized that the stem code 7100 can be augmented by a 4-value extension code that indicates which of the more specific functions is being evaluated. For this extended stem code, we used three qualifiers: the first indicates the specific joint that is involved (hip, in this case), the second indicates the laterality, and the third indicates severity, where the severity still depends on the specific function being evaluated.

Our detailed investigation of DBQ data elements suggested that developing a specific coding scheme from the outset was a suboptimal approach. First, a coding scheme is a syntactic construct and the optimal syntax is often dependent on specific use cases. For example, DBQs are organized in terms of data elements (e.g., "deep tendon reflexes of right knee") and data values (e.g., values from a 5-valued scale). From the point of view of capturing DBQ data, it was necessary to formulate the data as consisting of a data-element description and an acquired value, instead of coding it as a stem code with qualifiers. The key is that, if we had a consistent *semantic model* of the data elements and values, we could serialize them in alternative, equivalent syntaxes and infer the equivalence of data encoded in the different syntaxes.

Similarly, instead of creating arbitrary extensions to ICF codes, we could express the information content of the extensions more effectively as part of a semantic model of the data element. We needed to distinguish between *measurements* of functions, where the entities being measured are often represented by external terminologies, and *assessments* that are abstractions conceptually closer to the notions that ICF codes are designed to represent. Logical Observation Identifier Names and Codes (LOINC), for example, have many ready-made codes for the measurements that are recorded in DBQs (such as extension, rotation, and flexion of various joints). DBQ assessments, such as impairment of movement of the back (thoracolumbar spine), can be mapped to ICF. In both cases, detailed coding required that we add additional qualifiers, such as whether a range of motion is measured after repetition of actions. Such qualifiers could be added as attributes in a semantic model of the data elements.

Given the difficulty of obtaining de-identified data for development purposes, and the impracticality of abstracting functional assessment information from directly narrative text, we consulted with our Scientific Advisory Board. We came to the conclusion that the best way forward would be to focus not on existing unstructured data and a syntactic coding scheme such as the one in the original project proposal, where functional assessment information is represented by a code stem and a set of code-specific qualifiers, but rather on developing a **framework for structuring and using functional assessment information** prospectively. This framework included ontologies that describe the semantics of functional and related data elements, their relationships to standard terminologies and classifications, models of data-collection instruments, and data models for structuring assessed functional assessment information. We implemented the framework as a collection of ontologies using the Protégé tool. See Task 6 for details of how the semantic model for functional assessment information is structured.

The Protégé tool with which we created the ontologies and data models has a feature to export the content as a collection of inter-related HTML pages (Figure 3). This feature allowed us to integrate documentation for the model as part of the ontology.

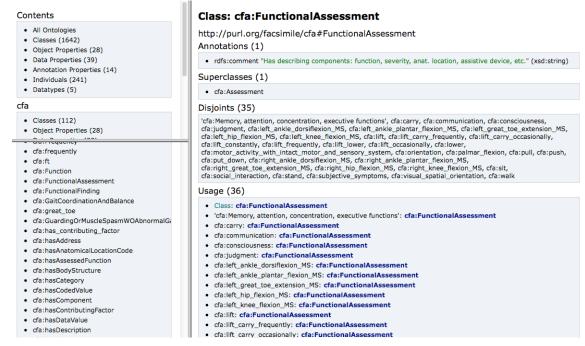


Figure 3. HTML pages documenting the CFA-CL ontology.

# 3. Using the proposed CFA-CL structure, we will develop a web-based editing tool for specifying CFA-CL code stems, their qualifiers, and value sets for the qualifiers.

We successfully imported the CFA-CL semantic model into WebProtégé, which provided us with a Web-based environment for editing the ontologies, data models, and service-member data (Figure 4).

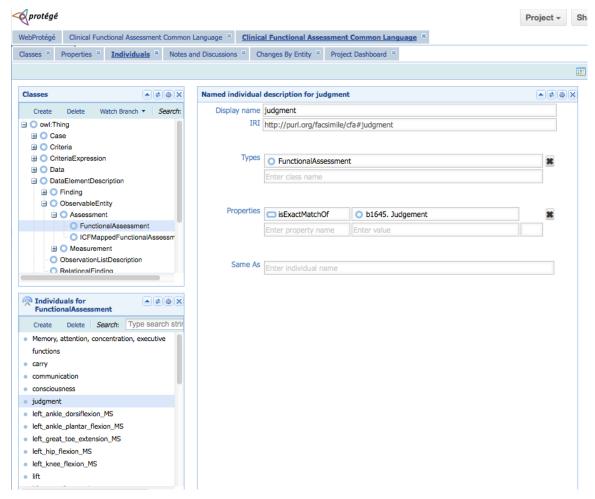


Figure 4. A WebProtege form for displaying and editing the "Judgment" data element. Note that this data element has an exact match to the ICF domain b1635.

4. We will populate CFA-CL with a selected subset of possible musculoskeletal code stems, their qualifiers, and value sets for the qualifiers.

We examined a set of existing instruments, including DBQs for lower back, knee and lower leg, ischemic diseases, and traumatic brain injury; the Military Occupation Specialties book; and SSA's residual functional assessments.<sup>2</sup> For the reasons discussed previously, we focused on the semantics of the associated functional entities and did not experiment with a specific coding syntax.

5. We will create a prototype CFA semantic model in which categories of impairment are defined by constraint expressions consisting of the CFA-CL and ICF code stems, qualifiers, and qualifier values.

In the CFA-CL framework, patient-specific functional-assessment data would be represented as semantic structures that are derived automatically as part of an enhanced data-entry process. We examined a set of existing instruments as described in Task 4 and modeled the structure and data elements in these instruments. Assessment instruments have sections and questions whose answers may be free text or may come from specific

<sup>&</sup>lt;sup>2</sup> The DBQs are VBA-21-0960M-14-ARE-Back.pdf, VBA-21-0960M-9-ARE-KneeLowerLeg.pdf, VBA-21-0960A-1-ARE-ischemic, NEURO - TBI Initial DBQ 9-15-11.doc.

value sets. Questions have descriptions of the data being solicited. Descriptions of questions and the value sets for their answers may use domain-specific terminologies.

To illustrate the structure of the CFA-CL Semantic Model, we take a question from the DBQ for the lower back. One of the assessments is a measurement of the forward flexion of the back (Figure 5):

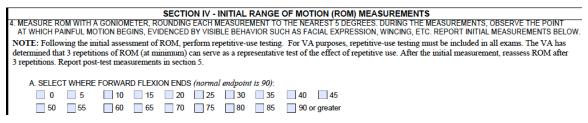


Figure 5. DBQ Range of Motion Measurement

We modeled the question as having an *isAbout* property (i.e., the data element description), text, and possible values (Figure 6):

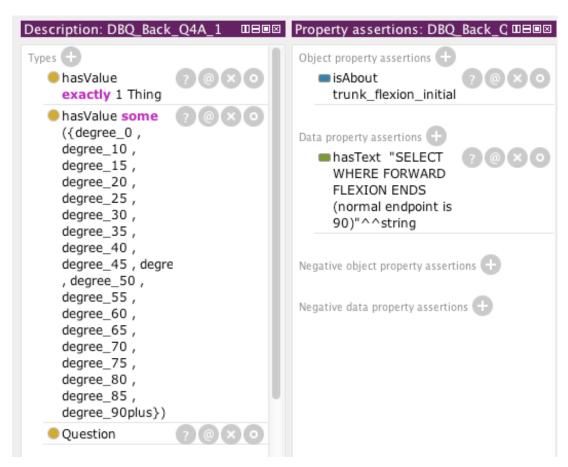


Figure 6. Modeling of Range of Motion data element.

The semantic model of the initial trunk-flexion data element was described in terms a number of properties (Figure 7):

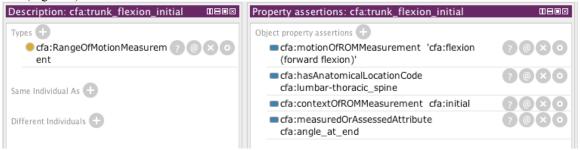


Figure 7. Semantic model for the trunk-flexion data element.

By associating the structured representation with components of an assessment instrument administered electronically, the acquired data could be converted as instances of the CFA-CL Semantic Model automatically, obviating the need to have human reviewers extracting and coding the data. A structured datum representing an initial trunk flexion measurement would look like an EHR datum (e.g., an Observation in the Health Level 7 Reference Information Model). At the minimum, it would have a reference to the focus of observation (e.g., trunk flexion initial), a value (e.g., 80 degrees), and the ID of the patient.

Our analysis of the data elements in assessment instruments suggested that multiple terminologies were needed to formalize the data-element descriptions. DBQs explicitly require the use of ICD for coding diagnoses. Many signs and symptoms are concepts better coded in standard clinical terminologies such as SNOMED CT. Among functional assessments, a significant subset involves detailed measurements such as assessments of the range of motion in specific joints. ICF, with its relatively high-level functional categories, is not designed for recording such measurements. We determined that, among standard clinical terminologies, LOINC has the appropriate codes for such measurements. For example, LOINC 41343-5 represents quantitative measurement of the angle of left-knee flexion. Currently, ICF is one of four standard terminologies to which we map descriptions of assessment-data elements. The mappings may be refined to specify that the data element description is an exact match, a specialization, or a generalization of the terminology concept.

# 6. We will define mappings between a selected subset of CFA-CL terms and ICF terms such that the mappings allow us to programmatically translate CFA-CL—coded data into corresponding ICF-coded data.

We modeled CFA-CL—coded data as instances of a class called datamodel:Observation (Figure 8). We investigated two alternative methods to relate such data, encoded in CFA-CL, to ICF. The first method, described here, translates the CFA-CL—coded data into ICF-coded data format, using Semantic Web Rule Language (SWRL) rules and a collection of mapping specifications encoded in the Web Ontology Language (OWL). The second method, described in Task 12, defined functional assessment data elements in terms of ICF concepts, making the resulting data queryable in terms of ICF concepts without converting the data into ICF-coded format.

## Use of SWRL rules to translate CFA-CL-coded data to ICF-coded data

For ICF-coded data, we create a model consisting of classes that corresponded to each of the four ICF axes: Body Structure, Body Function, Activities and Participation, and Environmental Factor. For each class, we specified the allowed qualifiers. Figure 9 shows the data model for "Body Structure" data. It specifies that an ICF-coded datum for body-structure impairment must include the nature, extent, and location of impairments.



Figure 8. Modeling of collected data as an Observation.



Figure 9. Model for "Body Structure" ICF-coded data.

To facilitate the use of Semantic Web Rule Language (SWRL) rules to perform the mapping from CFA-CL to ICF, we first created a mapping structure CFA2ICFMapping (Figure 10), where a CFA entity (e.g., an assessment term or a qualifier term) is mapped to the corresponding ICF category or qualifier value.

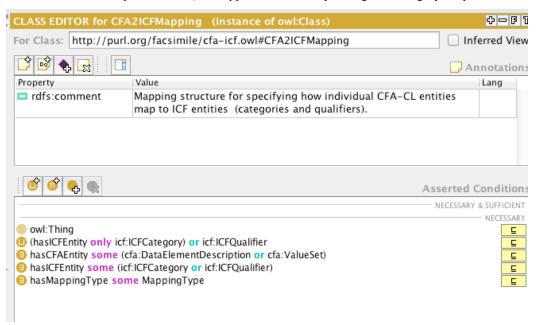


Figure 10. Mapping structure for translating CFA-CL coded data to ICF-coded data.

We modeled the mapping from the CFA-CL data format to the ICF data format as a collection of SWRL rules. An example is shown in Figure 11. It takes an Observation instance that encodes a body-function assessment

(e.g., right knee extension of 5 degrees) and mappings from CFA-CL to ICF (e.g., the notion of *extension* to "b7100 Mobility of a single joint") and that of 5 degrees to "3. SEVERE impairment (high, extreme, ...) 50–95 %" to create an instance of ICFBodyFunctionCode that denotes the mapped values as the combination of the ICF category and the 'extent of impairment' qualifier. To create the new ICF-coded data, we used Protégé's SWRL extension built-in swrlx:makeOWLIndividual, which is not one of the standard SWRL built-ins.

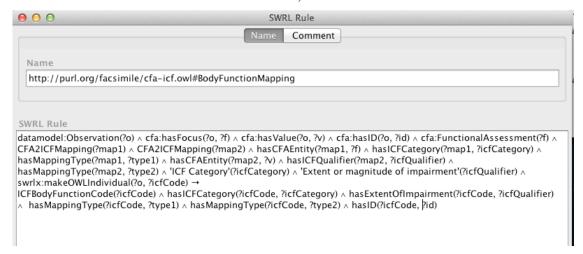


Figure 11. A SWRL rule for creating ICF-coded data from CFA-CL-coded body-function data.

Because ICF uses multiple codes to represent a single disability, we needed to write additional rules to translate the CFA-CL data to a set of ICF codes. For the example of "right knee extension" observation, we used a second rule to generate the ICF body structure code (Figure 12). Note that we used an *observation ID* to indicate that the ICF body function and body structure codes are derived from the same CFA-CL observation.

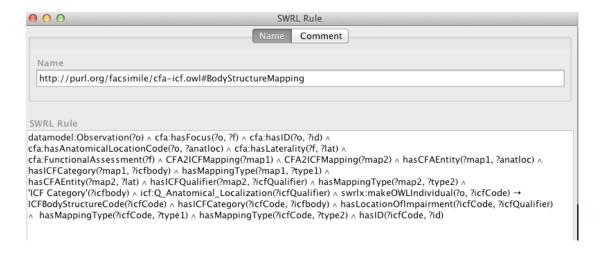


Figure 12. A SWRL rule to map the anatomical location of a body function assessment to ICF code.

7. We will create a developmental de-identified data set that contains musculoskeletal functional assessment. We will code the functional assessments using CFA-CL, and translate them to ICF-coded data.

As detailed in our report for Task 1, it was impossible to create de-identified data sets from the Madigan Army Medical Center archive. Instead, we developed the CFA-CL for the possibility of capturing data *prospectively* and we did not rely on the availability of de-identified data retrospectively.

# 8. We will define a set of queries that are interesting from the perspectives of evaluating individuals and of performing aggregated analysis. We will demonstrate the ability to make these queries on the developmental data set.

From our interviews at Madigan AMC, we came to the conclusion that Madigan providers are intensely focused on the evaluation of individual service members, and have little interest in queries of aggregated data.

Given our focus current focus on the DBQs, the queries that are most interesting from the perspective of evaluating individuals involve criteria from the Schedule for Rating Disabilities used in IDES to determine a numeric disability rating for the purpose of calculating the disability benefit. The criteria in the Rating Schedule are closely tied to questions in the DBQs. With our modeling of DBQ questions, we were able to answer such queries. For example, page 398 of the Schedule for Rating Disabilities states that, for an 10 percent disability rating, the service member should have, among possible alternatives,

muscle spasm, guarding, or localized tenderness not resulting in abnormal gait or abnormal spinal contour;

These criteria are directly related to questions in the example DBQ for the back (thoracolumbar spine) (Figure 13). In the CAF-CL ontology, we have a data element description for the finding *Guarding or Muscle Spasm of the Thoracolumbar Spine*, and, if a clinician populates the DBQ form generated from this project, the result is coded data of which the focus is this data element.

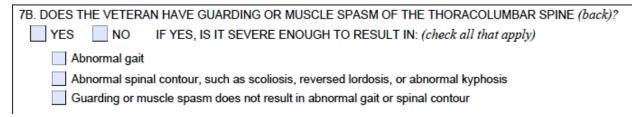


Figure 13. A question in the DQB on the back (thoracolumbar spine) conditions

With data coded in the CFA-CL Semantic Model that makes use of ICF concepts, we can make aggregated queries such as *most common disabilities associated with ICF code s7501 (structure of lower leg)*. We can aggregate disabilities to any level and sort by frequency. With these queries, we can identify the prevalence of specific problems (e.g., foot problems) that can be ameliorated with better equipment (e.g., change shoes, different inserts).

In Task 15, we demonstrated the ability to make such queries.

If we link CFA-CL data with other data sets, we can perform much more interesting queries. For example, with appropriate data sets, we can mine for associations between functional assessments and the risk of homelessness after discharge or between amputation and incidence of diabetes. We can identify the need for home support (e.g., the need for aid and attendance) based on functional losses. These possibilities indicate the potential for using structured functional assessment data, but creating such data sets is outside the scope of this project.

## 9. We will specify the IDES task for which we will demonstrate the use of CFA-CL—coded data.

We analyzed the criteria in the retention and rating standards. While many of these criteria, such as those related to range of motion, can be matched precisely from structured data, others, such as "Loss of toes that precludes

the abilities to run or walk without a perceptible limp and to engage in fairly strenuous jobs" require subjective judgment to identify. We may not be able to match such criteria with the data collected through any assessment instruments. Therefore, we believe that at most we can index the criteria with relevant codes, and that we can use the coded data for an individual subject, once the data become available, to focus attention on those criteria that may be relevant to that subject.

Given the unavailability of structured CFA data, we focused on the task of developing methods to acquire EHR-compatible structured data through assessment forms. Our modeling of components of assessment forms is similar to LOINC's approach, including the division into sections and questions, and the definition of possible answers to the questions. However, our work extends the LOINC representation by modeling the semantic content of the questions by giving these questions formal definitions in terms of a Clinical Functional Assessment (CFA) ontology, represented in OWL. The CFA ontology provides concepts and relationships that allow us to give formal descriptions of the findings, assessments, and measurements embodied in the assessment instruments. From the model of assessment instruments, we generate Web-based data-acquisition forms, through which clinicians can easily document necessary assessments. The backend of our tool automatically generates structured data in multiple formats. The data can be post-processed into formats consistent with those of Health Level 7 via simple transformations, and made available for querying and aggregation.

## 10. We will complete CFA-CL V0.5.

We define CFA-CL 0.5 as a framework for defining the semantic content of the assessment questions and answers by giving these questions and answers formal definitions in terms of a Clinical Functional Assessment (CFA) ontology. The CFA ontology provides concepts and relationships that allow us to give formal descriptions of the findings, assessments, and measurements embodied in the assessment instruments. In addition, we developed information models for such instruments and for data captured in the instruments. The CFA ontology and information models inform the generation of data-acquisition forms and the resulting data can be queried and aggregated. Our ontologies reference the ICF and other reference terminologies such as SNOMED CT. In order to allow generalization of this framework to other clinical domains, we created separate ontology files that can be re-used independently. In our specific application we use the full import closure as depicted in Figure 14. See appended paper "Structured Data Acquisition with Ontology-Based Web Forms" for detailed descriptions of the ontologies. Here we briefly describe the structure of the CFA ontology.

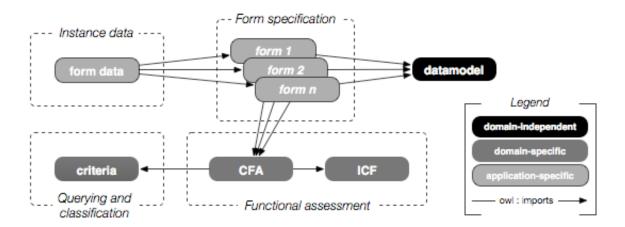


Figure 14. Import structure of ontologies developed as part of CFA-CL and its application to structured data acquisition through generated forms.

The CFA ontology is divided into three main branches: (1) DataElementDescription that defines a Finding (the result of an observation, measurement, or judgment, (2) ValueSet that defines collections of possible qualifiers and values for findings, and (3) SubjectMatterOntology that provides internally defined domain concepts that

either are not available in standard terminologies or are references to standard terms that need to be organized into taxonomies. The *Finding* class is further subdivided into *Assessment* (those findings that have non-numeric results) and *Measurement* (those findings that have numeric results). We also define *FunctionalAssessment* (a subclass of *Assessment*). In general, a functional assessment will have some assessed function that can be related to an ICF body function or activity (possibly as an exact match, specialization, or generalization), some assessed attribute, such as severity, that specifies the dimension of the function being assessed, and, optionally, some anatomical location of the assessment. Findings and functions can be modified by qualifiers that further refine these entities. For example, a functional assessment may be made in the context of using assistive devices, and a function being assessed may have some temporal component (e.g., constant pain). CFA imports a version of ICF that is represented in OWL. Thus, all ICF categories, such as 'body structure', 'body function', 'activities and participation', and 'environmental factors' are available for formalizing descriptions of functional assessments. For other standard terminologies such as SNOMED CT, ICD, and LOINC, instead of importing them as ontologies, we make references to them through instances of *ExternallyCodedValue*.

## 11. We will obtain de-identified data for demonstration purpose.

As detailed in our report for Task 1, it is impossible to create de-identified data sets from the Madigan Army Medical Center archive. We developed the CFA-CL for the possibility of capturing data prospectively and we are not relying on the availability of de-identified data retrospectively.

## 12. We will model CFA-CL V0.5 as parameterized constraints on ICF semantic model

ICF concepts are organized into components such as *body structure*, *body function*, *activity and participation*, and *environmental factors*. We integrate ICF concepts directly into descriptions of data elements. First, we use ICF's body structures as our default source ontology for anatomical locations. Second, we specify how the function being assessed is related to ICF functions using the properties *isExactMatchOf*, *isGeneralizationOf*, and *isSpecializationOf*. Third, we represent ICF qualifiers as either attributes being assessed (e.g., severity) or as qualifiers that modify the meaning of the functional assessment description (e.g., *laterality* modifying the anatomical location of the function being assessed). For example, "severity of constant pain on the lower left extremity cased by radiculopathy" would be modeled as shown in Figure 15.

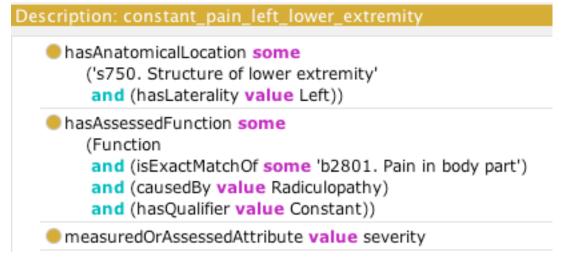


Figure 15. Modeling of the data element severity of constant pain on the lower left extremity cased by radiculopathy".

## 13. We will convert CFA-CL V0.5 codes to ICF coding format.

In Task 6, we showed a SWRL mapping approach to convert CFA-CL to ICF coding format. This approach has several disadvantages. First it requires that we come up with mapping functions, such as those that convert data-element—specific severity scales to the one used in the ICF coding scheme, that are difficult to justify. Second,

translating data elements into ICF-coded format is feasible only for functional assessment data, and thus such a translation places ICF-specific and non-ICF data into different data models, complicating queries that may join functional and non-functional assessment data. Non-ICF descriptors (e.g., attributes such as "intermittent" or "constant" that are used to qualify functional assessments cannot be represented in the ICF coding format. Finally, the use of SWRL rules to translate data from one format to another requires the use of non-standard extensions of SWRL that is available only in earlier versions of the Protégé tool that we use. For this milestone, we explored an alternative approach, as described in Task 12, where the mapping to ICF, when appropriate, is directly specified in the description of data elements (Figure 8).

In this approach, CFA-CL data are represented as instances of *Observations with a focus* (a reference to the CFA-CL description of a data element) and a value (e.g., see Figure 8). The reference to the CFA-CL description of a data element allows us to make detailed queries, as is described in Task 15.

Not translating data into pure ICF-coded format means that we cannot aggregate data across multiple data elements using ICF-imposed uniform qualifier values. Given the problems with such mappings (e.g., from degrees of extensions or rotations to a uniform 0-4 scale representing 'no impairment' to 'complete impairment 96-100%), such aggregation is probably not very meaningful anyway.

## 14. We will develop software for the application of CFA-CL V0.5 data to demonstration tasks .

See Appendix "Structured Data Acquisition with Ontology-Based Web Forms."

## 15. We will apply and analyze the application of CFA-CL V0.5 to the demonstration task.

See Appendix "Structured Data Acquisition with Ontology-Based Web Forms."

## 16. We will write the final report and package software, model artifacts, and sample data

## 4. Key Research Accomplishments

- We have come to the conclusion that current documentation practices in centers such as Madigan AMC pose difficulties in codifying clinical functional assessments as structured data.
- We have identified as a problem a lack of structured functional assessment data because there is no standard data representation that is in use. Yet the representation we are creating is difficult to evaluate because of the lack of data. A strategy to break the chicken-and-egg problem of data representation and data capture is to instrument systems for entering form-based data so that, as data are entered into forms, they are automatically transformed into our underlying models.
- We found that DBQs, the criteria in the MOS Manual and in the military retention standards require very specific functional assessments, which are difficult to map to ICF. There is no Rosetta Stone for translating neatly among these different data elements. What we can accomplish is to create a set of models that provides a mechanism for representing diverse data related to functional assessment.
- We found that, when the goal is to automate the capture of structured functional assessment data, the particular syntax that we initially proposed is not necessary. Instead the data can be structured in a semantically sound representation that facilitates queries and transformations.
- For the goal of capturing structured clinical functional assessment, we have created a semantic model of data element descriptions and a framework for using these descriptions to structure data.

- The modeling contributions include (1) CFA: a clinical functional assessment domain ontology that allows defining questions being asked in an assessment instrument in terms of a rich ontology that integrates standard terminologies such as ICF and SNOMED CT, and which provides the means for making detailed or aggregate queries on acquired data, and (2) a data model: an information model that allows the specification of generic assessment forms and the format of structured data acquired through the instruments. We have designed our output model to support the acquisition of structured data through Web forms, and for the potential to integrate the data inside EHRs. It is straightforward to transform the data we capture as instances of Observation, Certification, EvaluatorInformation, and SubjectInformation into, for example, Health Level Seven (HL7) Reference Information Model (RIM) standard compliant data.
- We developed the technology to generate forms for acquiring structured data based on the models forms, questions, and CFA CL data elements. The aggregated input gathered through these forms can be exported to databases and queried using standard SQL or can be represented an ontology of "semantically-enriched" form data that can be queried using an RDF query language, such as SPARQL

## 5. Conclusion

We have created a semantic framework for modeling structured functional-assessment data and showed how such data can be derived from assessment instruments such as DBQs. We have created mapping structures and rules to transform data represented in this framework to ICF-coded format.

We demonstrated (1) how to generate forms and acquire data based on the ontologies and data models in this semantic framework, and (2) how to make use of the data using queries on individual subjects and queries that aggregate population data.

## 6. Publications, Abstracts, and Presentations

Peer-Reviewed Conference Papers:

- [1] Gonc alves RS, Tu SW, Nyulas CI, Tieney MJ, Musen MA. Structured data acquisition with ontology-based web forms. International Conference on Biomedical Ontology. Lisbon, Portugal, 2015, in press. (Appendix I)
- [2] Tieney MJ, Tu SW, Nyulas CI, Gonc, alves\_RS, Musen MA. Driving structured data entry for functional assessment using standard terminologies. AMIA Annual Conference. San Francisco, CA, 2015, submitted. (Appendix II)

Abstracts:

[3] Musen MA, Nyulas CI, Tudorache T, Tu SW. Activities of the Stanford University WHO Collaborating Center. WHO - Family of International Classification Network Meeting. Barcelona, Spain 2014.

## 7. Inventions, Patents, and Licenses

Nothing to report

## 8. Reportable Outcomes

We have created a semantic model for clinical functional assessment consisting of

- An ontology of functional assessment data element descriptions
- An information model of assessment instruments and its components
- A data model for assessment data, in CFA-CL and ICF formats

We developed a mechanism that employs non-obtrusive ontology-based Web-forms to encode key functional assessment data, using terms from ICF and other standard terminologies. This solution allows us to query and aggregate the resulting structured data, based on standardized descriptions of assessment data elements. Our solution can advance adoption of standard terminologies, facilitate health information exchange and clinical decision support, and bring to bear the full power of modern electronic health records.

## 9. Other Achievements

Nothing to report

## 10. References

None.

## 11. Participants

Mark A. Musen MD, PhD Samson W. Tu, MS Csongor Nyulas, MS

## 12. Appendices

## Structured Data Acquisition with Ontology-Based Web Forms

Rafael S. Gonçalves, Samson W. Tu, Csongor I. Nyulas, Michael J. Tierney and Mark A. Musen

> Stanford Center for Biomedical Informatics Research Stanford University, Stanford, California, USA

#### **ABSTRACT**

Structured data acquisition is a common, challenging task that is widely performed in the field of biomedicine. However, in some biomedical fields, such as clinical functional assessment, little effort has been done to structure functional assessment data in such a way that it can be automatically employed in decision making (e.g., determining eligibility for disability benefits) based on conclusions derived from acquired data (e.g., assessment of impaired motor function). In order to be able to apply such automatisms, we need data structured in a way that can be exploited by automated deduction systems, for instance, in the Web Ontology Language (OWL); the de facto ontology language for the Web. The rise of OWL caused a paradigm shift in knowledge systems from frame-based to axiombased. Because of the axiom-based nature of OWL, it is more difficult to acquire instance data based on OWL than it was based on frames. In this paper we tackle the problem of generating Web forms from OWL ontologies, and aggregating input gathered through these forms as an ontology of "semantically-enriched" form data that can be queried using an RDF query language, such as SPARQL. The ontology-based structured data acquisition framework that we have developed is presented through its specific application to the clinical functional assessment domain, with examples of how one can perform desirable analyses of gathered data with simple queries.

#### 1 INTRODUCTION

Ontology-based form generation and structured data acquisition was first pioneered almost 30 years ago. In the early 1990s, Protégé-Frames used definitions of classes in an ontology to generate knowledge-acquisition forms, which could be used to acquire instances of the classes [2, 3]. With OWL as the preferred modeling language for ontologies, class definitions are collections of description logic (DL) axioms, and can no longer be seen as templates for forms [9]. Unlike template-based knowledge representations, where what can be said about a class is defined by the slots of the class template, axiom-based representations do not have this kind of locally scoped specification, and allow any axiom describing the same class to be added to the ontology, as long as the axiom does not lead to inconsistencies. Template-based knowledge representation systems use closed-world reasoning and have local constraints (e.g., cardinality of a slot for a particular class) that can be validated easily, while in an axiom-based system with the open-world assumption such local constraint checking is much more problematic. Furthermore, in our chosen application domain, assessment instruments have specific formats that do not lend themselves to be seen as representing instances of domain ontology classes. Items in the instruments have potentially complex

\*To whom correspondence should be addressed: rafaelsg@stanford.edu

descriptions of information to be collected, such as the severity of pain with a particular quality, and at a specific anatomical location. The challenge is to model the assessment instruments and relate the assessed data to a domain ontology with which one can formulate meaningful queries.

In this paper, we describe a solution for representing, acquiring and querying assessment data that uses (1) domain ontologies and standard terminologies to give formal descriptions of entities in our chosen domain, (2) an information model of assessment instruments to drive the generation of data-acquisition Web forms, and (3) a data model for the acquired information that links the data to the domain ontologies and standard terminologies. Such linkage makes it possible to query and aggregate the data using the logical representation of the domain concepts in the ontologies.

## 2 RELATED WORK

In addition to the comparison with Protégé-Frames' template-based instance acquisition method described in Section 1, we briefly contrast our work with two other systems that are designed to use forms for acquiring structured data: the first targets the domain of patient assessment, which is similar to the work reported here, while the second is a generic Web-based technology from which one can draw examples on how to arrive at a domain-independent solution.

The clinical documentation system described in [6] uses a template schema to allow a technology-savvy clinician to create documentation templates that include the local structure of subforms and potentially complex clinical descriptions consisting of features and their values. The features and values are mapped to a medical ontology, and the system automatically generates ontological descriptions of the data elements based on the mappings. Constrained by our goal to replicate existing forms, we took the opposite approach where we start with ontological descriptions of the data elements, specify how they are used in assessment instruments as part of the description of instruments, and generate Web forms for the acquisition of data. Having the freedom to design their documentation system, Horridge *et al.* avoided the laborious work of manually modeling the domain concepts.

Semantic wikis extend regular wikis with semantic technologies, wherein each wiki article is an RDF resource, and an instance of some resource such as a class defined in the schema, which can be asserted to have relations with other RDF resources. These relations are defined by the authors of wiki articles, which could be a challenging task to perform without previous knowledge of the domain or the modeling. In a survey of semantic wikis featuring OWL reasoning and SPARQL<sup>2</sup> querying facilities [4], a user

 $<sup>^{\</sup>rm 1}\,$  The typical kinds of schema accepted are OWL and RDFS.

http://www.w3.org/TR/rdf-sparql-query

evaluation of a chosen semantic wiki implementation concluded that authoring instance data in such a way is cumbersome, even with users that were familiar with ontologies. A good solution to this would be exploiting the relations defined in the schema to provide "wiki article templates" whose form input fields derive from those relations, thus making it easier to author semantic wiki articles.

## 3 APPLICATION DOMAIN

Clinical functional assessment provides the application motivation for our work. Functional assessment is the evaluation of an individual's ability to perform body functions (e.g., flexing a joint) and defined tasks (e.g., walking a specific distance). It is necessary for evaluating disabilities for rehabilitation, for social security payment, or for decisions to retain or discharge service members who may be injured on duty. Despite its importance, it is not usually supported by electronic health record (EHR) systems [1]. These assessments are often documented using assessment instruments (e.g., check-lists and validated questionnaires) such as Karnofsky Performance Status [11]. Too frequently the data derived from using these instruments are saved as either blobs or non-standard data elements. While a standard such as LOINC® (Logical Observation Identifiers Names and Codes) defines the syntactic structures of assessment instruments as a hierarchy of panels with questions that have coded answers [10], it does not relate the semantic content of the questions and answers to standard terminologies and data models that allow meaningful querying and aggregation of acquired data.

In our application scenario we use, as exemplars, the U.S. Department of Veterans Affairs (VA) Disability Benefits Questionnaires (DBQs). DBQs are used to evaluate service members' disabilities and to determine the benefits for which they are eligible. We start off with these DBQs as our initial form specifications, and design an ontology-based method for Web form generation and structured data acquisition, subsequently exemplifying how one would go about exploiting such data for immediate or *post facto* analyses.

## 4 MODELING

In order to capture the semantic distinctions that are needed in functional assessment, we developed a Clinical Functional Assessment (CFA) ontology that models the concepts and relationships that occur in functional assessment instruments. We developed information models for such instruments and for data captured in the instruments. We will show how the CFA ontology and information models inform the generation of data-acquisition forms and how the resulting data can be queried and aggregated. Our goal was to develop a set of light-weight ontologies and models with minimal ontological commitments, and postponing alignment with possible upper-level ontologies to the future. Existing ontologies, such as the Information Artifact Ontology (IAO),<sup>3</sup> do not provide a modeling of forms and questions that we could reuse. Furthermore, what we need is an information model that states, for example, that the structure of a "question" includes a specific text, not an ontology that models parts of information artifacts as ontological entities (e.g., modeling the text of a question as an instance of "textual entity" class). Our ontologies reference the

International Classification of Functioning, Disability and Health (ICF), <sup>4</sup> developed by the World Health Organization (WHO), and other reference terminologies such as SNOMED CT. <sup>5</sup>

**Imports structure** The modeling tasks of this project involve describing different domain areas, leading us to create separate ontology files that can be re-used independently. In our specific application we use the full import closure as depicted in Figure 1.

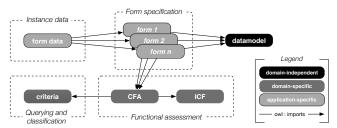


Fig. 1: Imports structure and role separation of ontologies developed for, or included as part of our modeling solution. Form specifications use terms from the *datamodel* ontology (e.g., to create question instances) as well as from domain-specific ontologies (e.g., CFA).

The ontology marked as Instance data in Figure 1 is the collection of data assertions from form submissions, possibly from different forms. The ontologies represented in Form specification are specifications of different forms; in our case, we use a single ontology that specifies two closely-related forms. The content of the above-mentioned ontologies is application-specific, that is, the way the data is represented is directly derived from the way in which forms are modeled (for different assessment instruments). However, resulting data still conform to the generic information models specified in the datamodel ontology. In this way, there is a separation of the Form specification ontologies (Abox axioms) from the Functional assessment ontologies that model the functional assessment domain and data models (mostly Tbox axioms). In Querying and classification we use a domain-specific ontology to apply SWRL rules,<sup>6</sup> and define complex OWL classes to facilitate querying in SPARQL and in OWL.

ICF ICF is a multi-purpose classification that, together with the International Classification of Diseases (ICD),<sup>7</sup> is a reference classification in the WHO Family of International Classifications (WHO–FIC). It provides a standard language and conceptual basis for the definition and measurement of functions and disability. However, unlike ICD codes that represent possible disease or injuries, coding different health and health-related states requires that ICF codes (e.g., "d4501" - walking long distance) be used in conjunction with component-specific qualifiers (e.g., a 0 to 4 scale to encode the range of impairment). Such a complex coding scheme makes it difficult to transform data derived from assessment instruments into the ICF format. Nevertheless, ICF provides a reference conceptual basis for the definition and measurement of functions and disability, thus justifying its usage in descriptions of functional assessment results, despite its limitations

 $<sup>\</sup>overline{^3}$  https://code.google.com/p/information-artifact-ontology

<sup>4</sup> http://www.who.int/classifications/icf/en

<sup>5</sup> http://www.ihtsdo.org/snomed-ct

<sup>6</sup> http://www.w3.org/Submission/SWRL

<sup>7</sup> http://www.who.int/classifications/icd/en

as a formal ontology [7]. To reference ICF concepts in our modeling of functional assessment descriptors, we use a version of ICF available from the National Center of Biomedical Ontology (NCBO) BioPortal repository [8], that is represented in OWL.

CFA The Clinical Functional Assessment (CFA) ontology models concepts and relationships that allow us to give formal descriptions of the findings, assessments, and measurements embodied in clinical functional assessment instruments. The ontology is divided into three main branches: (1) Finding: the result of an observation or judgement, (2) Value that defines collections of possible qualifiers and values for findings, and (3) SubjectMatterOntology that provides internally defined domain concepts that either are not available from standard terminologies or are references to standard terms that need to be organized into taxonomies. The Finding class is further subdivided into Assessment (those findings that have non-numeric result) and Measurement (those findings that have numeric results). We also define FunctionalFinding (a subclass of Finding) and Functional Assessment (a subclass of Assessment). In general, a functional assessment will have some assessed function that can be related to an ICF body function or activity (possibly as an exact match, specialization, or generalization), some assessed attribute, such as severity, that specifies the dimension of the function being assessed, and optionally some anatomical location of the assessment. Both findings and functions can be modified by qualifiers that further refine these entities. For example, a functional assessment may be made in the context of using assistive devices, and a function being assessed may have some temporal component (e.g., constant or intermittent pain). ICF being an imported ontology for CFA, all ICF categories, such as body structure, body function, activities and participation, and environmental factors are available for formalizing descriptions of functional assessments. For other standard terminologies such as SNOMED CT, ICD, and LOINC, instead of importing them as ontologies, we make references to them through an ExternallyCodedValue that specifies the terminology source and code. Queries that reference these codes require the availability of terminology services that relate these codes to other terms in the referenced terminologies.

The modeling of *Finding* is exemplified as follows, based on the "Back (Thoracolumbar Spine) Conditions" DBQ that we use as one of our exemplar assessment instruments; in the question on the severity of constant pain caused by radiculopathy on the right lower extremity, we define a subclass of *FunctionalAssessment* that has the assessed attribute 'severity', the assessed function 'icf:b2801 Pain in body part' that is qualified by a temporal quality 'Constant', and has anatomical location 'icf:s750. structure of lower extremity' with laterality 'Right'. Figure 2 illustrates the modeling of this assessment. With the modeling of the dimensions of assessment instrument questions, we can make queries on, and aggregate data collected through the instruments, as will be shown in Section 6.

```
    cfa:hasAnatomicalLocation some
        ('icf:s750. Structure of lower extremity'
        and (cfa:hasLaterality value cfa:Right))
    cfa:hasAssessedFunction some
        (cfa:Function
        and (cfa:isExactMatchOf some 'icf:b2801. Pain in body part')
        and (cfa:causedBy value cfa:Radiculopathy)
        and (cfa:hasQualifier value cfa:Constant))
    cfa:SeverityOfPainSensationCausedByRadiculopathy
```

Fig. 2: Modeling of "severity of constant pain caused by radiculopathy in the lower right extremity".

Datamodel The datamodel ontology is a generic, context-free representation of a form (e.g., it models elements such as questions and sections) and the data generated from a form (e.g., a string value from a text area, or values from an enumerated value set). Figure 3 summarizes key aspects of our modeling: elements of a form are asserted as subclasses of FormStructure, such as Form, Section and Question. Each kind of FormStructure generates some kind of Data; every form submission generates an instance of FormData, which references (via the hasComponent property) all instances of Data generated in the process of parsing form answers. Specific sections such as SubjectInfoSection collect information pertaining to a subject, and these details are aggregated in an instance of SubjectInformation. An answer to an instance of Question gives rise to an instance of Observation with a hasValue property assertion to the IRI of the selected answer. An instance of Observation will be inferred to have an outgoing hasFocus property assertion if the Question instance it derives from encodes some kind of semantic description of the question's meaning via the isAbout relation. Each instance of Question specifies a set of possible (answer) values via a hasPossibleValue relation to a subclass of Value.

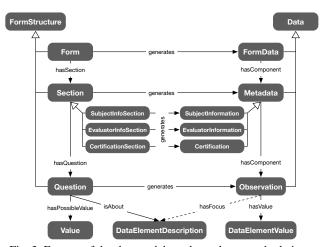


Fig. 3: Excerpt of the *datamodel* ontology classes and relations.

Form The Form ontology contains the set of individuals that are necessary to produce forms. While the technology we have developed is completely generic, we use as exemplars the U.S. Department of Veterans Affairs (VA) DBQs, which we modeled in an ontology named DBQ. This ontology contains instances of Question, Section, Form and other elements defined in the datamodel ontology (shown in Figure 3). Not only does this ontology rely on datamodel (for form structuring purposes), it also relies on functional assessment classes and individuals given in the CFA ontology, for example, values of a scale of severity of pain that should be presented as answer options to users reporting on the severity of constant pain in the lower extremity.

**Criteria** The *criteria* ontology contains SWRL rules to enrich the domain representation (e.g., if a *Question* instance has an *isAbout* relation with some instance i, then the *Observation* data instance that represents the answer to that question will get a *hasFocus* property filler i), as well as defined classes used to better support querying, which we describe in more detail in Section 6.

#### 5 OWL-BASED DATA ACQUISITION

Our approach to data acquisition in OWL requires two components: firstly, an OWL representation (in the form of one or more ontologies) of the form structures (questions, sections, etc), and descriptions of those structures' meanings, and, secondly, the view component that is given by an XML file specifying user-interface aspects. So, in order to use our method, a user will have to model questions and their descriptions in OWL, and then specify the layout and content of the resulting form in XML.

We implemented our form generation and data acquisition tool in Java, using the OWL API v4.0.1, <sup>8</sup> and its source code is publicly available on GitHub. <sup>9</sup> The tool implementation and configuration details are omitted here due to lack of space, but can be found in the GitHub project wiki. The tool takes as input a user-defined XML configuration file, generates a form, and outputs form answers in CSV, RDF and OWL formats. The configuration file should contain a pointer to the ontology specifying the form, as well as its imports. The two major stages in the service are form generation and form input handling, as described below.

- (1) Form generation Steps to produce a form:
  - (a) Process XML configuration, gathering form layout information, IRIs and bindings to ontology entities
  - (b) Extract from the input ontology all relevant information pertaining to each form element:
    - (b.1) Text to be displayed (e.g., section header, question text)
    - (b.2) Options and their text, where applicable
    - (b.3) The focus of each question
  - (c) Generate the appropriate HTML and JavaScript code
- (2) Form input handling Once the form is filled in and submitted:
  - (a) Process answer data and create appropriate individuals
  - (b) Produce a partonomy of the individuals created in (2.a) that mirrors the layout structure given in the configuration
  - (c) Return the (structured) answers to the user in a chosen format

The user-defined XML configuration (1.a) specifies: input and output information of the tool, bindings to ontology entities, and layout of form elements. The key XML elements are:

input: contains an ontology child element, and optionally a child element named imports

- ontology: absolute path or URL to the form specification ontology (e.g., DBQ ontology)
- *imports*: contains *ontology* child elements, which have an attribute *iri*, giving the IRI of the imported ontology

output: contains the following child elements

- file: defines, via a title attribute, the title of the form.
   Optionally, a path can be specified within the file element where the HTML form file should be serialized
- cssStyle: the CSS style class to be used in the output HTML bindings: defines mappings to ontology entities, such as what data property is used to state the text of a question, or section headings form: defines the layout and behaviors of the form

There is a wide range of versatility when configuring forms, such as: multiple levels of sub-questions, form element numbering,

question type (e.g., radio, checkbox, dropdown, horizontal checkbox, etc), question-list layout (vertical or inline) and recurrence; one can specify that a collection of questions should be repeated any given number of times. Some more complex options include overriding the default (alphabetic) order of answer options, and triggering sub-questions when a specific answer is selected. These two features are exemplified in Figure 4: this question is configured with an attribute/value pair: showSubquestionsForAnswer="cfa:Yes" on the question XML element, so that answering 'Yes' triggers the sub-questions of that question. In Figure 4, under 'Right lower extremity', we have a question with a list of answer options derived from an enumerated value set, which would ordinarily be ordered alphabetically. However, 'None' would then appear between 'Moderate' and 'Severe', thus interrupting a severity scale. So we added: optionOrder="3;\*" to the question element, which states that the would-be third option (alphabetically) should appear first, and the remaining (the "\*" wild character stands for "all unmentioned options") should be presented in default order.



Fig. 4: The user interface of the form generated for the DBQ question corresponding to radiculopathy pain modeled in Figure 2.

The key output of the data acquisition tool is the OWL ontology, as it provides us with "semantically enriched" form data that can be used for aggregation and querying. The resulting data individuals are structured in OWL (via hasComponent relations) similarly to how the form is structured in the configuration, that is, if question Q is configured as having two sub-questions, then the Observation individual generated by Q will have two outgoing hasComponent relations to the instances of Observation generated by the two sub-questions of Q.

## 6 DATA ANALYSIS

One of the authors (Michael J. Tierney), who is a physician from the VA Palo Alto Healthcare System, validated the generated OWL-based versions of the DBQ forms, and filled in the "Back (Thoracolumbar Spine) Conditions" DBQ with 5 complete sets of sample data. The data gathered are stored in a graph database with support for SPARQL 1.1 querying and OWL 2 reasoning.

Since our data are both structured and semantically enriched, we are able to query the observations using SPARQL, classify them into criteria representing powerful OWL expressions, or manipulate them using SWRL. For example, Code Snippet 1 presents a simple SPARQL query that returns all instances of *Observation* where a patient presented signs or symptoms due to radiculopathy. It is worth observing that this query is formulated in such a way that it is independent of the assessment instrument, including the particular formulation of the question, but rather uses the appropriate focus individual from our CFA ontology.

<sup>8</sup> http://owlapi.sourceforge.net

 $<sup>^{9}</sup>$  http://github.com/protegeproject/facsimile

**Code Snippet 1** SPARQL query for retrieving all observations of radicular pain due to radiculopathy.

```
SELECT ?obs WHERE {
    ?obs a datamodel:Observation .
    ?obs datamodel:isDerivedFrom ?q .
    ?q a datamodel:Question .
    ?q cfa:isAbout
        cfa:signs_or_symptoms_due_to_radiculopathy .
    ?obs cfa:hasValue cfa:Yes }
```

In order to query for all observations of severe pain anywhere in the lower extremity, one could formulate an OWL DL query such as that given in Code Snippet 2.

**Code Snippet 2** OWL DL query for retrieving all observations of severe pain anywhere in the lower extremity.

In response to the query in Code Snippet 2, a DL reasoner uses the semantic descriptions of the observation foci, which are derived from the questions' *isAbout* property, to aggregate answers for severe pain for different parts of the lower extremity.

## 7 DISCUSSION

In this paper we presented a framework for OWL-based form generation and data acquisition that gathers form answers as tabdelimited data, RDF triples, or OWL instances, which can be subsequently analyzed in a systematic way (as shown in our queries in Section 6). Once the raw data is processed (by deriving the foci of observations from the *isAbout* field of the questions), the resulting data have no dependency on specific questions (except for provenance tracking), so if the form specification is modified, then previous form data are still comprehensible and sound (i.e., upon form specification changes the new data and old data remain compatible). However, if a user requires data to be structured in a different or more specialized format than ours, then either the software needs modifying, or a post-processing step would be necessary. The value of data in such a structured format in any arbitrary domain is twofold: automating, or improving the automation of the process of arriving at desirable conclusions from questions in the form, and for further analysis, for instance, via querying. In the clinical functional assessment domain, our modeling of forms and questions is consistent with the format of assessment instruments defined in LOINC. However, the types of queries we formulated for functional assessment data are unfeasible using LOINC, since LOINC provides no semantics behind what an answer to a specific question means.

We presented our modeling of functional assessments and assessment instruments, and demonstrated (1) how to generate forms and acquire data based on these OWL ontologies and data models, and (2) how to make use of the data using queries on individual subjects and queries that aggregate population data.

The modeling contributions include (1) CFA: a clinical functional assessment domain ontology that allows defining questions being asked in an assessment instrument in terms of a rich ontology that integrates standard terminologies such as ICF and SNOMED CT, and which provides the means for making detailed or aggregate queries on acquired data, and (2) datamodel: an information model that allows the specification of generic assessment forms and the format of structured data acquired through the instruments.

We have designed our output model to support the acquisition of structured data through Web forms, and for the potential to integrate the data inside EHRs. It is straightforward to transform the data we capture as instances of *Observation*, *Certification*, *EvaluatorInformation*, and *SubjectInformation* into, for example, Health Level Seven (HL7) Reference Information Model (RIM) standard compliant data [5]. Finally, we have shown that the problem of structured data acquisition can be suitably tackled using OWL; our solution, though applied to the clinical functional assessment domain for the context of this paper, is entirely generic, and can easily be applied to an arbitrary domain.

## **ACKNOWLEDGMENTS**

This work is supported in part by contract W81XWH-13-2-0010 from the U.S. Department of Defense, and grants GM086587 and GM103316 from the U.S. National Institutes of Health (NIH).

#### **REFERENCES**

- [1] Buyl, R. and Nyssen, M. (2009). Structured electronic physiotherapy records. *Int. J. of Med. Inf.*, **78**(7), 473–481.
- [2] Eriksson, H., Puerta, A. R., and Musen, M. A. (1994). Generation of knowledge-acquisition tools from domain ontologies. *Int. J. of Human-Computer Studies*, 41, 425–453.
- [3] Gennari, J. H., Musen, M. A., Fergerson, R. W., Grosso, W. E., Crubzy, M., et al. (2003). The evolution of Protégé: an environment for knowledge-based systems development. Int. J. of Human-Computer Studies, 58(1), 89–123.
- [4] Gonçalves, R. S. (2009). Semantic Wiki for Travel and Holidays using OWL. Master's thesis, The University of Manchester.
- [5] Health Level Seven (2015). HL7 Reference Information Model. www.hl7.org/implement/standards/rim.cfm.
- [6] Horridge, M., Brandt, S., Parsia, B., and Rector, A. (2014). A domain specific ontology authoring environment for a clinical documentation system. In *Proc. of CBMS-14*.
- [7] Kumar, A. and Smith, B. (2005). The ontology of processes and functions: A study of the international classification of functioning, disability and health. In *Proc. of the AIME Workshop* on *Biomedical Ontology Engineering*.
- [8] Noy, N. F., Shah, N. H., Whetzel, P. L., Dai, B., Dorf, M., et al. (2009). BioPortal: ontologies and integrated data resources at the click of a mouse. *Nucleic Acids Research*, 37, 170–173.
- [9] Rector, A. (2013). Axioms & templates: Distinctions & transformations amongst ontologies, frames & information models. In *Proc. of K-CAP-13*.
- [10] Vreeman, D. J., McDonald, C. J., and Huff, S. M. (2010). Representing patient assessments in LOINC(R). In *Proc. of AMIA*.
- [11] Yates, J. W., Chalmer, B., McKegney, F. P., et al. (1980). Evaluation of patients with advanced cancer using the Karnofsky performance status. CANCER, 45(8), 2220–2224.

# Driving Structured Data Entry for Functional Assessment Using Standard Terminologies

Michael J. Tierney, MD<sup>1,2</sup> Samson W. Tu, MS, <sup>1</sup> Csongor I. Nyulas, MS, <sup>1</sup>
Rafael S. Gonçalves PhD, <sup>1</sup> Mark A. Musen MD, PhD<sup>1</sup>

Stanford Center for Biomedical Informatics Research, Stanford University,

Stanford, CA, USA

<sup>2</sup>VA Palo Alto Health Care System, Palo Alto, CA

## **ABSTRACT**

Since its introduction in 2001, the International Classification of Functioning, Disability and Health (ICF) has matured into a recognized standard for describing human functioning in the context of illness. Many have adopted instruments to help incorporate use of the complex terminology into research and clinical documentation. Yet it remains unclear how this standard can be best utilized in an electronic health record (EHR) in order to take advantage of the benefits such a standard provides, while improving usability for clinicians. We developed a mechanism that uses ontology-based Web-forms to insulate the user from the details of ICF coding. The resulting form data are linked to logical descriptions that use terms from ICF and other standard terminologies. This solution allows us to query and aggregate the resulting structured data based on standardized descriptions of assessment data elements, to advance adoption of standard terminologies in clinical functional assessment.

## INTRODUCTION

In the spring of 2001, the World Health Organization (WHO) introduced the International Classification of Functioning, Disability and Health (ICF) to describe non-fatal health outcomes, and to define functioning and patient progress in rehabilitation, with special attention to the role of the environment in human functioning <sup>1, 2, 3</sup>. It offers researchers and policymakers a uniform standard language for describing and discussing disability<sup>4</sup>, as well as assessing need for services and resources<sup>5</sup>. The ICF consists of broad multi-dimensional assessments using qualifiers and numeric coding schemes, and, over time, many have apportioned and focused the terminology to specific disciplines and fields to further its utility in clinical and research settings<sup>2</sup>.

Despite its status as the companion classification of the International Classification of Diseases (ICD) in the WHO's Family of International Classifications (WHO-FIC), the adoption of ICF in clinical functional assessment is modest. In this paper, we survey the literature to explore issues related to the adoption and uptake of ICF, especially in the context of its usage to derive machine-usable structured data. This survey informs us of the requirements that any solution that advances the use of standard terminologies in clinical functional assessment should meet. Using Semantic Web technologies, particularly the Web Ontology Language (OWL), we propose a new method to incorporate ICF in a clinical functional assessment (CFA) ontology that provides the basis for defining data elements in structured assessment instruments. Using formal models of assessment forms, we developed a prototype application that automatically generates Web-based data-entry forms. Through these forms, a healthcare provider can document clinical functional assessment and generate EHR-compatible structured data that can be used for decision support or population-based queries.

## BACKGROUND

The WHO built the ICF as a hierarchical classification of 1,424 coded categories and 1,122 definitions<sup>6</sup> divided primarily into two parts: 'Functioning and Disability', and 'Contextual Factors', and further subdivided into four components: 'Body Functions and Structures', 'Activities and Participation', 'Environmental Factors', and 'Personal Factors' (see Figure 1). Each ICF code is assigned one or more numeric qualifiers to reflect the level of facilitation or impairment conferred by a health condition<sup>5</sup>.

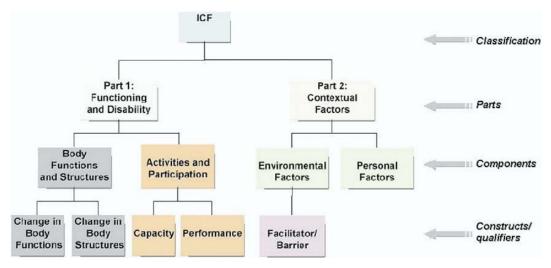


Figure 1. Hierarchy of the ICF, depicting its major division in two parts, and including components and qualifiers (reused with permission from Kukafka et al 2006, originally adapted from WHO, 2006).

## Clinical Challenges to Use of ICF

Despite many advancements in incorporating ICF into the research and clinical realms <sup>1, 2, 6, 7, 8</sup>, challenges remain in the clinical setting to implement and utilize the ICF during patient visits. The broad scope of the ICF is felt as potentially overwhelming or impractical for clinicians to use in a given clinical encounter<sup>3,6</sup>, while at the same time lacking some codes that were sufficiently unique or detailed<sup>3,9</sup>. Critics pointed to lack of guidance on clinical implementation, resulting in uneven and idiosyncratic implementation<sup>3, 8</sup>, and others cited lack of empirical evidence of clinical utility and questioned whether the complex implementation of ICF had any benefit for patients at all<sup>8</sup>. Anner et al felt that the application of ICF to describe work disability was limited since it does not include key features of the disability evaluation, such as the dynamic time perspective or the restricted causal connection between functional capacity and the health condition<sup>6</sup>. Perhaps as a result, ICF coding has achieved limited integration into clinical documentation in physiotherapy<sup>10</sup>. This suggests that ICF needs to be complemented with other standard terminologies, and appropriate uses of the terminology should be predefined so that they are implemented correctly, yet details are hidden from the user.

#### **Outcomes Research and Linking Rules**

"Linking Rules" were soon developed and refined to promote the application of ICF as a connecting framework between interventions and outcome measures, and to allow researchers to systematically link and compare meaningful concepts contained in different health-status measures, technical and clinical measures, and interventions<sup>11</sup>. The use of ICF as a reference standard for patient-oriented outcome measures is practical when such a standardized procedure enables interventions and outcome measures to be linked to the ICF terminology<sup>4</sup>. Several authors have found that linking rules have practical value in clinical settings<sup>3,12,13,14</sup>.

## The Electronic Health Record (EHR)

Incorporating physiotherapy functional assessment in a coded format into electronic health records can help to leverage features that are valuable for health information exchange, clinical decision support, and can potentially improve patient care outcomes<sup>10</sup>. Vreeman and Richoz describe how the ICF could advance clinical practice and research by enabling data sharing and reuse by EHRs<sup>10</sup>. However, incorporating coded functional assessment into an EHR faces some hindrances. Beyond ICF, there are several alternative standards and assessments available to clinicians, for example: Discipline-specific, SNOMED, LOINC, etc.

Documentation formats range from paper notes, free text entry, and structured data entry, all of which raise unique challenges for clinicians and computers regarding how the data is recorded and analyzed. Any solution to EHR incorporation of ICF-based assessments should address the variety of standards and assessment instruments, and take into account the preferred documentation format.

## Structured Data Entry

Since unstructured 'free text' narrative, commonly seen in rehabilitation assessment notes, is not easily utilized by software, some have advocated Natural Language Processing (NLP) approaches to improving coding of ICF with assessment data entry. Kukafka and her colleagues advocate the practicality of the automated Medical Language Extraction and Encoding system (MedLEE) NLP system, which selects ICF codes and performance qualifiers better than non-expert human coders, though not as well as expert human coders. They found that the NLP system produced levels of agreement on code assignments that were generally higher for the first qualifier (performance) and lower for the second qualifier (capacity). However, coding tasks involving complex reasoning (i.e., requiring assimilation of multiple sources and types of information as might be required in a disability assessment) remain a difficult challenge for current NLP systems<sup>5</sup>.

Because of these difficulties with NLP systems, others have advocated the advantages of structured data entry. Structured data entry offers a viable pathway in implementing and using ICF within the EHR. Vreeman and Richoz propose template-based data entry to facilitate data collection and analysis by computer, framed by ICF standards<sup>10</sup>. They describe the incorporation of ICF coding into an EHR, yielding specific clinical reminders for providers to address, as well as enhancing abilities to generate reports, calculate function scores, and enhance clinical decision making. While many clinicians balk at structured data entry in the clinical encounter and can find coding terminology distracting from the flow of patient records, Vreeman and Richoz suggest that the identifiers (codes) and names from the vocabulary can be hidden inside the EHR while the clinician interacts with customary concept labels<sup>10</sup>. They call for additional work to define the relationships between assessment instruments, ICF and other vocabulary standards in a computer-interpretable way<sup>10</sup>.

In recent years, LOINC has been extended to capture data elements in assessment instruments<sup>15</sup>. LOINC allows the aggregation of assessment items into a hierarchy of panels. For each assessment item, LOINC maintains attributes such as 'Question Text', 'Question Source', data type of answers, and structured answer lists. Much of our representation of assessment forms, questions, and answers parallel LOINC's organization. LOINC, however, does not attempt to model the questions and answers in assessment instruments semantically in terms of ontologies relevant to functional assessments. Each LOINC assessment item stands alone. For example, a visual assessment item for "near acuity" has no relationship to another item "far acuity". Because these two assessment items are not logically related (in a hierarchy), one cannot use automated reasoning to infer that a subject has a generalized visual acuity problem based on responses to the two assessment items.

### **METHODS**

Given the requirements we have outlined in the previous section, we focus on developing methods to acquire EHR-compatible structured data through assessment forms. Additionally, we explore the use of Semantic Web technologies to develop new types of linking rules for systematically linking and comparing meaningful concepts contained in different health-status measures, technical and clinical measures, and interventions. Our modeling of components of assessment forms is similar to LOINC's approach, including the division into sections and questions, and the definition of possible answers to the questions. However, our work extends the LOINC representation by modeling the semantic content of the questions by giving these questions formal definitions in terms of a Clinical Functional Assessment (CFA) ontology, represented in OWL. The CFA ontology provides concepts and relationships that allow us to give formal descriptions of the findings, assessments, and measurements embodied in the assessment instruments. From the model of assessment instruments, we generate Web-based data-acquisition forms, through which clinicians can easily document necessary assessments. The backend of our tool automatically generates structured data in multiple formats. The data can be post-processed into formats consistent with those of Health Level 7 via simple transformations, and made available for querying and aggregation.

Clinicians use the "Back (Thoracolumbar Spine) Conditions" Disability Benefits Questionnaire (DBQ) to assess veteran's musculoskeletal disability. We use this DBQ as one of our exemplar assessment instruments (see Figure 2) with the objective of capturing, as structured data, answers documented by these clinicians. We will illustrate the use of the CFA ontology to structure the information about the severity of constant pain caused by radiculopathy of the left lower extremity.

SECTION XII - RADICULOPATHY							
12A. DOES THE VETERAN HAVE RADICULAR PAIN OR ANY OTHER SIGNS OR SYMPTOMS DUE TO RADICULOPATHY?							
YES NO IF YES, COMPLETE THE FOLLOWING SECTION:							
12B. INDICATE SYMPTOMS' LOCATION AND SEVERITY (check all that apply):							
Constant pain (may be excruciating at times)							
Right lower extremity: None Mild Moderate Severe							
Left lower extremity: None Mild Moderate Severe							

Figure 2. A fragment of the "Back (Thoracolumbar Spine) Conditions" Disability Benefits Questionnaire. It shows the dependency of questions (question 12B needs to be completed only if the answer to question to 12A is "YES") and how a question (e.g., 12B) is subdivided into subquestions (severity of pain in either lower extremity).

The CFA ontology is divided into three main branches: (1) DataElementDescription that defines a Finding (the result of an observation, measurement, or judgment, (2) ValueSet that defines collections of possible qualifiers and values for findings, and (3) SubjectMatterOntology that provides internally defined domain concepts that either are not available in standard terminologies or are references to standard terms that need to be organized into taxonomies. The Finding class is further subdivided into Assessment (those findings that have non-numeric results) and Measurement (those findings that have numeric results). We also define Functional Assessment (a subclass of Assessment). In general, a functional assessment will have some assessed function that can be related to an ICF body function or activity (possibly as an exact match. specialization, or generalization), some assessed attribute, such as severity, that specifies the dimension of the function being assessed, and, optionally, some anatomical location of the assessment. Findings and functions can be modified by qualifiers that further refine these entities. For example, a functional assessment may be made in the context of using assistive devices, and a function being assessed may have some temporal component (e.g., constant pain). CFA imports a version of ICF that is represented in OWL. Thus, all ICF categories, such as 'body structure', 'body function', 'activities and participation', and 'environmental factors' are available for formalizing descriptions of functional assessments. For other standard terminologies such as SNOMED CT, ICD, and LOINC, instead of importing them as ontologies, we make references to them through instances of Externally Coded Value.

Figure 3 shows how the assessment of constant pain caused by radiculopathy in the left lower extremity is modeled. To make it easier for descriptions of questions and answers in a form to reference this class, we create an individual that is the "prototypical instance" of the class (as shown in ).

```
    cfa:hasAnatomicalLocation some
        ('icf:s750. Structure of lower extremity'
        and (cfa:hasLaterality value cfa:Left))
    cfa:hasAssessedFunction some
        (cfa:Function
        and (cfa:isExactMatchOf some 'icf:b2801. Pain in body part')
        and (cfa:causedBy value cfa:Radiculopathy)
        and (cfa:hasQualifier value cfa:Constant))
    cfa:measuredOrAssessedAttribute value cfa:severity
```

Figure 3. Definition of constant pain caused by radiculopathy on the lower left extremity. It shows that the finding is defined in terms of the function being assessed, the assessed attribute of the function, and the

anatomical location of the assessment. In each case, the value of the property can be constrained by expressions that reference internally defined value sets or terms from external terminologies.

Besides the CFA ontology, we have also created a datamodel ontology, which provides a generic, context-free representation of a form (e.g., it models elements such as questions and sections) and the data generated from a form (e.g., a string value from a text area, or values from an enumerated value set). Figure 4 summarizes key aspects of our modeling: elements of a form are asserted as subclasses of *FormStructure*, such as *Form*, *Section* and *Question*. Each kind of *FormStructure* generates some kind of *Data*; every form submission generates an instance of *FormData*, which references (via the *hasComponent* property) all instances of *Data* generated in the process of parsing form answers. Specific sections such as *SubjectInfoSection* collect information pertaining to a subject, and these details are aggregated in an instance of *SubjectInformation*. An answer to an instance of *Question* gives rise to an instance of *Observation* with a *hasValue* property assertion to the IRI of the selected answer. An instance of *Observation* will be inferred to have an outgoing *hasFocus* property assertion if the question it derives from encodes some kind of semantic description of the question's meaning. Figure 5. shows the modeling of a question about constant pain caused by radiculopathy of the lower left extremity.

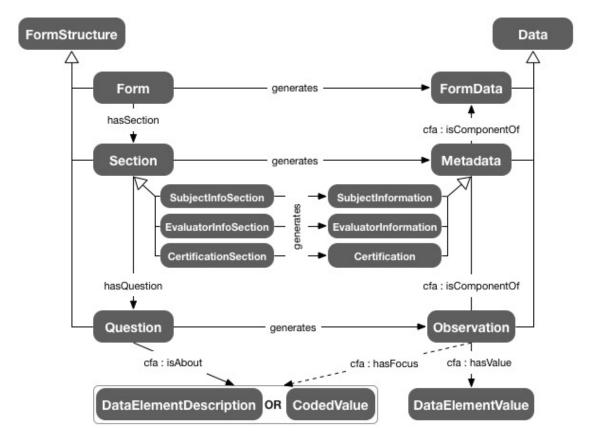


Figure 4. Excerpt of the datamodel ontology classes and relations. The left-hand side of the figure shows the components of a form and how a question's semantic content (the value of the cfa: *isAbout* property) is defined by concepts in the CFA ontology. The right-hand side of the figure shows the structure of the data generated from answers to these questions.



Figure 5. Modeling of a question about constant pain caused by radiculopathy of the lower left extremity. The *cfa:isAbout* property references an individual (*constant\_pain\_left\_lower\_extremity*) in the CFA ontology that is the prototypical instance of a class of the same name. The *cfa:hasText* property specifies the string that should be displayed with the question, and the *cfa:hasValue* property is constrained by the value set for answers to this question.

Based on the modeling of forms and questions, we developed a Java-based application that generates a Web form that a healthcare provider can use to enter assessments that are captured as structured data by the backend (see Figure 6). The application takes as input a specification of a form (such as the 'Back' DBQ form) in an ontology, and a user defined configuration file.

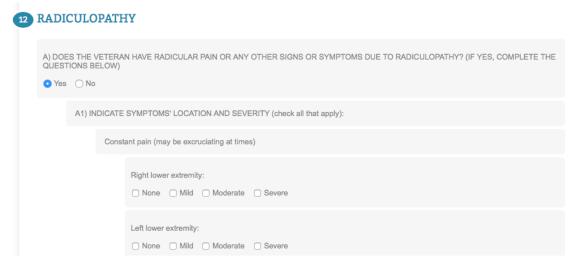


Figure 6. DBQ questions concerning the symptoms of radiculopathy. The form was generated from a specification of the questions on the "Back (Thoracolumbar Spine) Conditions" DBQ. After a healthcare provider submits assessment data using this form, the system generates structured data using the form specification, the output data model, and concepts from the CFA ontology.

## RESULTS

We have developed a proof-of-concept system that includes complete modeling of two DBQs. One of the authors (MJT), who is a physician from the VA Palo Alto Healthcare System, reviewed and validated the generated Web forms, and created data for a number of simulated patient cases. The data generated can be saved as tab-delimited files or Semantic Web documents (RDF or OWL). While the data can be transformed into formats consistent with those of HL7 data format, we stored our gathered data in a graph database with support for SPARQL 1.1 querying and OWL 2 reasoning.

The data captured through our Web forms is both structured and semantically enriched because it is linked to the CFA ontology's modeling of data elements. We create a link by asserting a question's semantic description, encoded as an *isAbout* relation, into the *hasFocus* property of the data derived from that question. For example, the back DBQ has a question about the severity of constant pain caused by radiculopathy of the left lower extremity. An observation derived from this question will have a focus that is a reference to the class defined by the expression shown in , and some value, for instance, 'severe'. This

kind of semantic modeling allows us to query and aggregate the data along the dimensions that define a clinical functional assessment (e.g., severity, assessed function, and anatomical location). Figure 7 shows an OWL query that retrieves all observations of severe pain anywhere in the lower extremity. It is worth observing that this query is formulated in such a way that it is independent of the assessment instrument, including the formulation of the question. Instead, the query is formulated in terms of entities defined in our CFA ontology.

```
datamodel:Observation and
cfa:hasValue value cfa:severe and
cfa:hasFocus some (cfa:Assessment and
  (cfa:hasAssessedFunction some(cfa:isExactMatchOf some icf:b2801)) and
  (cfa:hasAnatomicalLocation some icf:s750))
```

Figure 7. OWL query for retrieving all observations of severe pain in the lower extremity.

#### DISCUSSION

We have demonstrated a method to bridge the gap between assessment instruments commonly used to perform clinical functional assessment and standard terminologies such as ICF. Our CFA ontology allows us to define questions in assessment instruments in terms of concepts and relationships specified in the ontology. These formal descriptions, in turn, reference terms from standard terminologies such as ICF and SNOMED CT. The descriptors of functional assessment—the function and its attribute being assessed, possible anatomical locations, and qualifiers on assessments and functions—are parallel to the categories and qualifiers of ICF. However, the value sets for these descriptors do not have to be limited to ICF, and can use terms from other standard terminologies. Furthermore, the ontology provides the flexibility of linking functional assessment concepts with other clinical entities. For example, in our example of pain caused by radiculopathy, we used the *caused by* property to link the pain being assessed to the SNOMED CT concept for radiculopathy. Users of the assessment instruments do not have to understand the complex ICF coding scheme and its extensions. The system automatically generates structured data consistent with the use of ICF.

The CFA ontology plays a role similar to that of "Linking Rules" described in the Background section. It provides a logic-based connecting framework for comparing and harmonizing intervention and outcome measures from different instruments. Because of the amount of work needed to build consensus and to formally describe the needed functional assessments, organizations that develop discipline-specific assessment instruments need to take the lead to define appropriate components of the CFA ontology.

Our prototype implementation of the form-based assessment instruments have some standard features that help to save time for healthcare providers, such as automated population of sub-questions if a top-level question has a certain value (e.g., "all normal"). Nevertheless an experienced clinician no doubt can document functional assessment more rapidly using a paper form. The benefit of using an instrument like ours is the possibility of automatically generating structured data that can be queried, aggregated, and transformed into standard formats, thus bringing clinical functional assessment one step closer to being integrated with a modern EHR.

## **CONCLUSION**

The adoption of ICF has been hindered by a structure that coders and clinicians find difficult to use. Ontology-based forms for structured data entry can offer a bridge from the clinical encounter to computerized data analysis. Ideal solutions should mesh well with clinical workflow and enable data recording by clinicians and researchers in a logical and rapid fashion. Such technical solutions should likewise be configurable to different terminologies to address the needs of clinicians and researchers. Structured data entry is a recognized, though imperfect, format for recording data in the clinical encounter, and has back-end advantages over NLP systems. We have built on existing terminologies and reuse their terms to construct an ontology of functional assessment. This ontology overcomes significant limitations of the native ICF coding structure.

We developed a mechanism that employs non-obtrusive ontology-based Web-forms to encode key functional assessment data, using terms from ICF and other standard terminologies. This solution allows us

to query and aggregate the resulting structured data, based on standardized descriptions of assessment data elements. Our solution can advance adoption of standard terminologies, facilitate health information exchange and clinical decision support, and bring to bear the full power of modern electronic health records.

## Acknowledgments

This work is supported by contract W81XWH-13-2-0010 from the U.S. Department of Defense and grants GM086587 and GM103316 from the U.S. National Institutes of Health (NIH).

#### References

- 1. Üstün TB, Chatterji S, Bickenbach J, Kostanjsek N, Schneider M. The International Classification of Functioning, Disability and Health: a new tool for understanding disability and health. Disabil Rehabil. 2003;25:565-571.
- 2. Stucki G, Cieza A, Ewert T, Kostanjsek N, Chatterji S, ÜstÜn TB. Application of the International Classification of Functioning, Disability and Health (ICF) in clinical practice. Disabil Rehabil. 2002;24:281-282.
- 3. Reed GM, Lux JB, Bufka LF et al. Operationalizing the International Classification of Functioning, Disability and Health in Clinical Settings. Rehabil Psychol. 2005;50:122.
- 4. Fayed N, Cieza A, Edmond Bickenbach J. Linking health and health-related information to the ICF: a systematic review of the literature from 2001 to 2008. Disabil Rehabil. 2011;33:1941-1951.
- 5. Kukafka R, Bales ME, Burkhardt A, Friedman C. Human and automated coding of rehabilitation discharge summaries according to the International Classification of Functioning, Disability, and Health. J Am Med Inform Assoc. 2006;13:508-515.
- 6. Anner J, Schwegler U, Kunz R, Trezzini B, de Boer W. Evaluation of work disability and the international classification of functioning, disability and health: what to expect and what not. BMC Public Health. 2012;12:470.
- 7. Ewert T, Fuessl M, Cieza A et al. Identification of the most common patient problems in patients with chronic conditions using the ICF checklist. J Rehabil Med. 200422-29.
- 8. Wiegand NM, Belting J, Fekete C, Gutenbrunner C, Reinhardt JD. All talk, no action?: the global diffusion and clinical implementation of the international classification of functioning, disability, and health. Am J Phys Med Rehabil. 2012;91:550-560.
- 9. Jelsma J. Use of the International Classification of Functioning, Disability and Health: a literature survey. J Rehabil Med. 2009;41:1-12.
- 10. Vreeman DJ, Richoz C. Possibilities and Implications of Using the ICF and Other Vocabulary Standards in Electronic Health Records. Physiother Res Int. 2013
- 11. Cieza A, Geyh S, Chatterji S, Kostanjsek N, Ustun B, Stucki G. ICF linking rules: an update based on lessons learned. J Rehabil Med. 2005;37:212-218.
- 12. Escorpizo R, Finger ME, Glässel A, Cieza A. An international expert survey on functioning in vocational rehabilitation using the international classification of functioning, disability and health. J Occup Rehabil. 2011;21:147-155.
- 13. Laxe S, Tschiesner U, Zasler N, Lopez-Blazquez R, Tormos JM, Bernabeu M. What domains of the International Classification of Functioning, Disability and Health are covered by the most commonly used measurement instruments in traumatic brain injury research? Clin Neurol Neurosurg. 2012;114:645-650.
- 14. Cerniauskaite M, Quintas RUI, Boldt C et al. Systematic literature review on ICF from 2001 to 2009: its use, implementation and operationalisation. Disabil Rehabil. 2011;33:281-309.
- 15. Vreeman DJ, McDonald CJ, Huff SM. Representing Patient Assessments in LOINC(R). AMIA Annu Symp Proc. 2010;2010:832-836.